PROTEIN CALCULATOR

Back to the Calculator Back to Work Pages

Keratin 14 (NP_000517)

Accepted Sequence

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NLNDRLASYL DKVRALEEAN ADLEVKIRDW YQRQRPAEIK DYSPYFKTIE DLRNKILTAT  180
VDNANVLLQI DNARLAADDF RTKYETELNL RMSVEADING LRRVLDELTL ARADLEMQIE  240
SLKEELAYLK KNHEEEMNAL RGQVGGDVNV EMDAAPGVDL SRILNEMRDQ YEKMAEKNRK  300
DAEEWFFTKT EELNREVATN SELVQSGKSE ISELRRTMQN LEIELQSQLS MKASLENSLE  360
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GEDAHLSSSQ FSSGSQSSRD VTSSSRQIRT KVMDVHDGKV VSTHEQVLRT KN          472
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Molecular Weight = 51624.59

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Last Updated: May 26, 1999

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LOCUS       NP_000517                472 aa            linear   PRI 20-DEC-2003
DEFINITION  keratin 14; cytokeratin 14 [Homo sapiens].
ACCESSION   NP_000517
VERSION     NP_000517.2  GI:15431310
DBSOURCE    REFSEQ: accession NM_000526.3
KEYWORDS    .
SOURCE      Homo sapiens (human)
  ORGANISM  Homo sapiens
            Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi;
            Mammalia; Eutheria; Primates; Catarrhini; Hominidae; Homo.
REFERENCE   1  (residues 1 to 472)
  AUTHORS   Schuilenga-Hut,P.H., Vlies,P., Jonkman,M.F., Waanders,E., Buys,C.H.
            and Scheffer,H.
  TITLE     Mutation analysis of the entire keratin 5 and 14 genes in patients
            with epidermolysis bullosa simplex and identification of novel
            mutations
  JOURNAL   Hum. Mutat. 21 (4), 447 (2003)
   PUBMED   12655565
  REMARK    GeneRIF: Three novel KRT14 mutations identified in 9 Epidermolysis
            bullosa simplex patients.
REFERENCE   2  (residues 1 to 472)
  AUTHORS   McGargill,M.A., Mayerova,D., Stefanski,H.E., Koehn,B., Parke,E.A.,
            Jameson,S.C., Panoskaltsis-Mortari,A. and Hogquist,K.A.
  TITLE     A spontaneous CD8 T cell-dependent autoimmune disease to an antigen
            expressed under the human keratin 14 promoter
  JOURNAL   J. Immunol. 169 (4), 2141-2147 (2002)
   PUBMED   12165543
  REMARK    GeneRIF: A spontaneous CD8 T cell-dependent autoimmune disease to
            an antigen expressed under the human keratin 14 promoter.
REFERENCE   3  (residues 1 to 472)
  AUTHORS   Chan,Y., Anton-Lamprecht,I., Yu,Q.C., Jackel,A., Zabel,B.,
            Ernst,J.P. and Fuchs,E.
  TITLE     A human keratin 14 'knockout': the absence of K14 leads to severe
            epidermolysis bullosa simplex and a function for an intermediate
            filament protein
  JOURNAL   Genes Dev. 8 (21), 2574-2587 (1994)
   PUBMED   7525408
REFERENCE   4  (residues 1 to 472)
  AUTHORS   Stephens,K., Sybert,V.P., Wijsman,E.M., Ehrlich,P. and Spencer,A.
  TITLE     A keratin 14 mutational hot spot for epidermolysis bullosa simplex,
            Dowling-Meara: implications for diagnosis
  JOURNAL   J. Invest. Dermatol. 101 (2), 240-243 (1993)
   PUBMED   7688405
REFERENCE   5  (residues 1 to 472)
  AUTHORS   Hovnanian,A., Pollack,E., Hilal,L., Rochat,A., Prost,C.,
            Barrandon,Y. and Goossens,M.
  TITLE     A missense mutation in the rod domain of keratin 14 associated with
            recessive epidermolysis bullosa simplex
  JOURNAL   Nat. Genet. 3 (4), 327-332 (1993)
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PUBMED 7526933
REFERENCE 6 (residues 1 to 472)
AUTHORS Bonifas,J.M., Rothman,A.L. and Epstein,E.H. Jr.
TITLE Epidermolysis bullosa simplex: evidence in two families for keratin gene abnormalities
JOURNAL Science 254 (5035), 1202-1205 (1991)
PUBMED 1720261
REFERENCE 7 (residues 1 to 472)
AUTHORS Coulombe,P.A., Hutton,M.E., Letai,A., Hebert,A., Paller,A.S. and Fuchs,E.
TITLE Point mutations in human keratin 14 genes of epidermolysis bullosa simplex patients: genetic and functional analyses
JOURNAL Cell 66 (6), 1301-1311 (1991)
PUBMED 1717157
REFERENCE 8 (residues 1 to 472)
AUTHORS Rosenberg,M., RayChaudhury,A., Shows,T.B., Le Beau,M.M. and Fuchs,E.
TITLE A group of type I keratin genes on human chromosome 17: characterization and expression
JOURNAL Mol. Cell. Biol. 8 (2), 722-736 (1988)
PUBMED 2451124
REFERENCE 9 (residues 1 to 472)
AUTHORS Albers,K. and Fuchs,E.
TITLE The expression of mutant epidermal keratin cDNAs transfected in simple epithelial and squamous cell carcinoma lines
JOURNAL J. Cell Biol. 105 (2), 791-806 (1987)
PUBMED 2442174
REFERENCE 10 (residues 1 to 472)
AUTHORS Marchuk,D., McCrohon,S. and Fuchs,E.
TITLE Complete sequence of a gene encoding a human type I keratin: sequences homologous to enhancer elements in the regulatory region of the gene
JOURNAL Proc. Natl. Acad. Sci. U.S.A. 82 (6), 1609-1613 (1985)
PUBMED 2580298
REFERENCE 11 (residues 1 to 472)
AUTHORS Marchuk,D., McCrohon,S. and Fuchs,E.
TITLE Remarkable conservation of structure among intermediate filament genes
JOURNAL Cell 39 (3 Pt 2), 491-498 (1984)
PUBMED 6210150
REFERENCE 12 (residues 1 to 472)
AUTHORS Hanukoglu,I. and Fuchs,E.
TITLE The cDNA sequence of a human epidermal keratin: divergence of sequence but conservation of structure among intermediate filament proteins
JOURNAL Cell 31 (1), 243-252 (1982)
PUBMED 6186381
COMMENT REVIEWED REFSEQ: This record has been curated by NCBI staff in collaboration with Michael Rogers. The reference sequence was derived from J00124.1.
On Sep 5, 2001 this sequence version replaced gi:4504913.

Summary: This gene encodes a member of the keratin family, the most diverse group of intermediate filaments. This gene product, a type I keratin, is usually found as a heterotetramer with two keratin 5 molecules, a type II keratin. Together they form the cytoskeleton of epithelial cells. Mutations in the genes for these keratins are associated with epidermolysis bullosa simplex. At least one pseudogene has been identified at 17p12-p11.
FEATURES Location/Qualifiers

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